

DNA STATISTICAL ANALYSIS FOR RELATIONSHIP TESTING

A. SCOPE

When individuals cannot be excluded as being biologically related, statistical calculations are performed to aid in understanding the strength of the association. The following procedure outlines the statistical calculations used during relationship testing.

A.1 PATERNITY CALCULATIONS:

Probability of Parentage (W) determines a posterior (i.e. after all evidence is considered) probability for parentage based upon the DNA profiles of a known parent, child and alleged parent.

$$W_{\text{locus}} = \frac{(PI_{\text{locus}}) * P}{(PI_{\text{locus}}) * P + (1-P)}$$

P is the prior probability. The prior probability of paternity is the strength of one's belief that the tested man is the father based only on the non-genetic evidence. The prior probability is given the neutral value of 0.5. That is, Popstats assumes there is a 50% probability that the alleged parent is the biological parent of the child and a 50% probability that the alleged parent is not the biological parent of the child.

PI is the parentage index (see below for explanation)

Looking at all loci:

$$W = \frac{CPI * P}{CPI * P + (1-P)}$$

CPI is the combined parentage index (see below for explanation)

Parentage Index (PI)

The parentage index is a likelihood ratio based on two conditional probabilities:

The probability of a child's genotype given that the alleged parent is the biological parent.

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The probability of a child's genotype given that the alleged parent is not the biological parent.

The general formula is as follows:

$$PI = \frac{\text{Probability of genetic observations conditional on parentage}}{\text{Probability of genetic observations conditional on non-parentage}}$$

The PI is calculated for each locus and then the individual PI values are multiplied together to obtain the Combined Paternity Index (CPI) for the entire set of genetic loci examined. A PI of 100 or greater correlates to the probability that the alleged father has a 99 to 1 better chance of being the father than a random man.

Probability of Exclusion (PE)

The probability of exclusion is defined as the probability of excluding a random individual from the population given the alleles of the child and the mother. The genetic information of the tested man is not considered in the determination of the probability of exclusion. The probability of exclusion is equal to the frequency of all men in the population who do not contain an allele that matches the obligate paternal allele of the child. The probability of Exclusion (PE) for a DNA profile is determined by the following equation:

$$1 - [(1 - PE_{\text{locus1}}) \times (1 - PE_{\text{locus2}}) \times (1 - PE_{\text{locus3}}) \dots \times (1 - PE_{\text{locusN}})]$$

Reverse Parentage

Reverse parentage statistics evaluate the likelihood that the DNA evidentiary sample is from the child of a pair of known biological parents.

Mutations

If a child and the tested individual do not match at three or more tested STR loci, then the tested individual is excluded as being the biological father/mother of the child.

If a child and the tested individual do not match at one or two STR locus/loci:

Y STR analysis should be performed where possible when the relationship of two males is in question.

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A paternity index must be calculated for the inconsistent locus/loci, which takes into account the possibility of a mutation. The mutation rate and mean power of exclusion for the locus can be entered using the Popstats program.

A mutation may be one of the possible explanations; the genetic results could suggest that a close relative (such as a brother, child or father) may be the biological father. Therefore, the avuncular index may be required to determine the likelihood that the tested individual is an uncle.

If a mutation is suspected, then the appropriate samples may be sent out to a paternity laboratory for mutational analysis.

Kinship analysis

Kinship evaluates the likelihood that the pair of given DNA profiles are associated by kinship versus by chance. Single parent or motherless paternity cases utilize kinship analysis. Refer to Popstats for relevant formulas. All other types of kinship statistical analysis are not performed in our laboratory at this time.

The formulas recommended by the American Association of Blood Banks will be used. Refer to "Parentage Testing Accreditation Requirement Manual, American Association of Blood Banks" Third Edition.

Linkage of loci vWA and D12S391

Due to the close proximity of vWA and D12S391, one of these two loci will be dropped in all paternity calculations performed using the GlobalFiler kit; only the more conservative locus will be utilized.

B. PROCEDURE: PARENTAGE TRIO

Using Popstats, the following is how to obtain the parentage index and the probability of parentage (paternity) when you have a known mother and child and an alleged father:

- B.1** Select the Parentage Statistics button and select the Trio Parentage option.
- B.2** Enter the biological mother's sample into the "Known Parent" column.
- B.3** Enter the Child's sample into the "Child" column.
- B.4** Enter the alleged father's sample into the "Alleged Parent" column.
- B.5** Click the calculate statistics button on the toolbar.

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- B.6** The parentage index indicates whether the evidence fits better with the hypothesis that the man is the father or with the hypothesis that someone else is the father. The probability of parentage is the measure of the strength in one's belief (non-genetic combined with genetic evidence) in the hypothesis that the tested man is the father.
- B.7** Put all the individual parentage indexes in the table that will be attached to the report. Note the combined parentage indexes on this page also.
- B.8** See DOC ID [1790](#) for report wording.

C. PROCEDURE: REVERSE PARENTAGE

Using Popstats, the following is how to obtain the reverse parentage index and the probability of exclusion when you have an unknown child and an alleged mother and father. For example, in the case of an abandoned baby the reverse parentage statistics can be used to evaluate if the abandoned baby is the biological child of the suspected couple:

- C.1** Select the Parentage Statistics button and select the Reverse Parentage option.
- C.2** Enter the suspected mother's profile into the Known Mother column.
- C.3** Enter the suspected father's profile into the Known Father column.
- C.4** Enter the questioned child's profile into the Alleged Child column.
- C.5** Click the calculate statistics button on the toolbar.
- C.6** The reverse parentage index indicates whether the evidence fits better with the hypothesis that the child is the biological child or with the hypothesis that someone else is the biological child. The probability of exclusion for reverse parentage is the probability of excluding a random person from the population as being the biological child given the alleles of the parents.
- C.7** Put all the individual parentage indexes in the table that will be attached to the report. Note the combined reverse parentage indexes on this page also.
- C.8** See DOC ID [1790](#) for report wording.

D. PROCEDURE: Single Parentage

Using Popstats, the following is how to obtain the single parentage index and the probability of exclusion for single parentage when you have an unknown child and an alleged mother / father.

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For example, in the case of an abandoned baby the single parentage statistics can be used to evaluate the possibility that the abandoned baby is the biological child of the suspected mother or father.

NOTE: Testing without the mother presents a number of problems. First, the paternity index value is, on average, cut in half. Additionally, when a mother's sample is not tested, the combined paternity index value is about one tenth of that observed when the mother's sample is tested. The absence of the mother's sample also greatly reduces the ability to detect a falsely accused man, and in some cases such as incest, can produce false inclusions. Thus, the testing of the mother, even if maternity is not disputed, is important in evaluating the questioned relationship; it improves the chance of obtaining clear results and is a quality control check for both the scientific and legal community. Testing without the mother should only be done when mother's location is unknown or she is deceased. Every effort should be made to test the mother:

D.1 Select Kinship Statistics button and select PO (parent-offspring) kinship option.

D.2 Enter the child's profile in the Reference column.

D.3 Enter the suspected mother's profile into the Evidence column.

D.4 Click the calculate statistics button on the toolbar.

D.5 The single parentage index indicates whether the evidence fits better with the hypothesis that the person is the biological parent or with the hypothesis that someone else is the biological parent. The probability of exclusion for single parentage is the probability of excluding a random person from the population as the biological parent given the alleles of the child. In other words, it is the frequency of all people in the population who do not contain an allele that matches the child.

D.6 Put all the individual parentage indexes in the table that will be attached to the report. Note the combined single parentage indexes on this page also.

D.7 See DOC ID [1790](#) for report wording.

E. PROCEDURE: Parentage Trio with a single or double mutation

Using Popstats, the following is how to obtain the parentage index and the probability of parentage (paternity) when you have a known mother and child and an alleged father. The alleged father does not match at one or two locations. This inconsistency cannot be ignored and a paternity index must be calculated for the inconsistent locus/loci, which takes into account the possibility of a mutation(s). Refer to the AABB's annual reports for information regarding mutation frequencies and the mean power of exclusion (PE). STRbase (<http://www.cstl.nist.gov/strbase/>) may also be useful.

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NOTE: The AABB standards indicate that laboratories may not exclude with one inconsistency without supporting evidence. The guidance document cautions laboratories that double or even triple mutation can occur. The laboratory should be cautious in issuing a report as exclusionary with only two inconsistencies. (Taken from the AABB Annual Summary Report for Testing, 2010).

- E.1** Select the Parentage Statistics button and select the Trio Parentage option.
- E.2** Enter the biological mother's sample into the "Known Parent" column.
- E.3** Enter the Child's sample into the "Child" column.
- E.4** Enter the alleged father's sample into the "Alleged Parent" column.
- E.5** Click the calculate statistics button on the toolbar.
- E.6** Enter the Paternal Mutation Frequency and the mean power of exclusion for the locus/loci containing the mutation(s); Popstats will automatically provide these numbers during the calculation.
- E.7** The parentage index indicates whether the evidence fits better with the hypothesis that the man is the father or with the hypothesis that someone else is the father. The probability of parentage is the measure of the strength in one's belief (non-genetic combined with genetic evidence) in the hypothesis that the tested man is the father.
- E.8** Put all the individual parentage indexes in the table that will be attached to the report. Note the combined parentage indexes on this page also.
- E.9** See DOC ID [1790](#) for report wording.

F. REFERENCES

- F.1** "Parentage Testing Accreditation Requirement Manual, American Association of Blood Banks" Third Edition.
- F.2** CODIS Training Manual
- F.3** <http://www.cstl.nist.gov/strbase/>
- F.4** <http://www.aabb.org/Pages/Homepage.aspx>

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F.5 http://www.cstl.nist.gov/strbase/pub_pres/NIST-D12-vWA-EDNAP-Apr2011.pdf

F.6 <http://www.aabb.org/sa/facilities/newsletter/Documents/relationship-testing-news-1405.pdf>

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